47,XYY syndrome

47,XYY syndrome is characterized by an extra copy of the Y chromosome in each of a male's cells. Although males with this condition may be taller than average, this chromosomal change typically causes no unusual physical features. Most males with 47,XYY syndrome have normal sexual development and are able to father children.

47,XYY syndrome is associated with an increased risk of learning disabilities and delayed development of speech and language skills. Delayed development of motor skills (such as sitting and walking), weak muscle tone (hypotonia), hand tremors or other involuntary movements (motor tics), and behavioral and emotional difficulties are also possible. These characteristics vary widely among affected boys and men.

A small percentage of males with 47,XYY syndrome are diagnosed with autistic spectrum disorders, which are developmental conditions that affect communication and social interaction.

Frequency

This condition occurs in about 1 in 1,000 newborn boys. Five to 10 boys with 47,XYY syndrome are born in the United States each day.

Genetic Changes

People normally have 46 chromosomes in each cell. Two of the 46 chromosomes, known as X and Y, are called sex chromosomes because they help determine whether a person will develop male or female sex characteristics. Females typically have two X chromosomes (46,XX), and males have one X chromosome and one Y chromosome (46,XY).

47,XYY syndrome is caused by the presence of an extra copy of the Y chromosome in each of a male's cells. As a result of the extra Y chromosome, each cell has a total of 47 chromosomes instead of the usual 46. It is unclear why an extra copy of the Y chromosome is associated with tall stature, learning problems, and other features in some boys and men.

Some males with 47,XYY syndrome have an extra Y chromosome in only some of their cells. This phenomenon is called 46,XY/47,XYY mosaicism.

Inheritance Pattern

Most cases of 47,XYY syndrome are not inherited. The chromosomal change usually occurs as a random event during the formation of sperm cells. An error in cell division called nondisjunction can result in sperm cells with an extra copy of the Y chromosome.

If one of these atypical reproductive cells contributes to the genetic makeup of a child, the child will have an extra Y chromosome in each of the body's cells.

46,XY/47,XYY mosaicism is also not inherited. It occurs as a random event during cell division in early embryonic development. As a result, some of an affected person's cells have one X chromosome and one Y chromosome (46,XY), and other cells have one X chromosome and two Y chromosomes (47,XYY).

Other Names for This Condition

- Jacob's syndrome
- XYY Karyotype
- XYY syndrome
- YY syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Double Y syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C3266843/

Other Diagnosis and Management Resources

- Association for X and Y Chromosome Variations: Tell Me About 47,XYY
  https://genetic.org/variations/about-xyy/

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Health Topic: Learning Disorders
  https://medlineplus.gov/learningdisorders.html
- Health Topic: Speech and Communication Disorders
  https://medlineplus.gov/speechandcommunicationdisorders.html

Genetic and Rare Diseases Information Center

- 47, XYY syndrome
  https://rarediseases.info.nih.gov/diseases/5674/47-xyy-syndrome

Educational Resources

- Disease InfoSearch: 47, XYY Syndrome
  http://www.diseaseinfosearch.org/47%2C+XYY+Syndrome/43
- MalaCards: 47,xyy syndrome
  http://www.malacards.org/card/47xyy_syndrome
- March of Dimes: Chromosomal Conditions
  http://www.marchofdimes.org/baby/chromosomal-conditions.aspx
- Merck Manual Consumer Version
  http://www.merckmanuals.com/home/children-s-health-issues/chromosomal-and-genetic-abnormalities/xyy-syndrome
- Orphanet: 47,XYY syndrome
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=8

Patient Support and Advocacy Resources

- Association for X and Y Chromosome Variations
  https://genetic.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/xyy-syndrome/
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/chromoso.html#xyy
- Unique: The Rare Chromosome Disorder Support Group (UK)
  http://www.rarechromo.org/information/Chromosome_Y/XYY%20FTNW.pdf

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%2247%2Cxyy+syndrome%22+OR+%22xyy+karyotype%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28xyy%5BTIAB%5D%29+OR+%28yy+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

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